GFM1 gene

G elongation factor mitochondrial 1

Normal Function

The *GFM1* gene provides instructions for making an enzyme called mitochondrial translation elongation factor G1. This enzyme is found in cell structures called mitochondria, which are the energy-producing centers within cells. While instructions for making most of the body's proteins are found in DNA that is stored in the nucleus of cells (nuclear DNA), a few proteins and other molecules are produced from DNA that is stored in mitochondria (mtDNA). Mitochondrial translation elongation factor G1 is involved in the production of proteins from mtDNA through a process called translation. The mtDNA genes provide instructions for products that are involved in protein production and the process of turning energy taken in from food into a form that cells can use (oxidative phosphorylation).

During translation, mtRNA molecules, which are the protein blueprints created from mtDNA, interact with specialized complexes called ribosomes to assemble protein building blocks (amino acids) into a fully formed protein. The role of mitochondrial translation elongation factor G1 is to coordinate the movements of mtRNA molecules with ribosomes to allow assembly of the protein to continue until it is complete.

Health Conditions Related to Genetic Changes

Combined oxidative phosphorylation deficiency 1

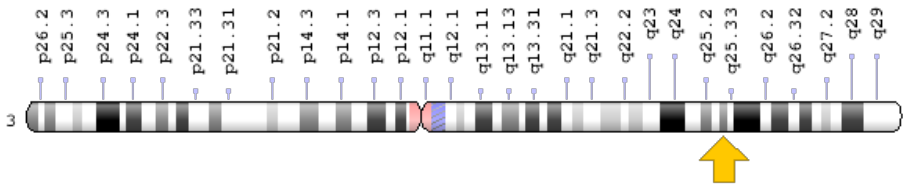
At least 18 mutations in the *GFM1* gene have been found to cause combined oxidative phosphorylation deficiency 1. This condition causes severe neurologic and liver dysfunction. Affected individuals usually do not survive past early childhood. Most of the *GFM1* gene mutations change single amino acids in the mitochondrial translation elongation factor G1 enzyme. Such alterations reduce or eliminate the enzyme's function. As a result, fewer mitochondrial proteins involved in oxidative phosphorylation are produced. Organs that have high energy demands, such as the brain and liver, are particularly affected by the resulting impairment of oxidative phosphorylation. A shortage of energy in these tissues leads to cell death, causing the neurological and liver problems in people with combined oxidative phosphorylation deficiency 1. It is thought that other tissues that require a lot of energy, such as the heart and other muscles, are not affected in this condition because they have additional enzymes that can perform the process of mitochondrial protein production.

Leigh syndrome

Chromosomal Location

Cytogenetic Location: 3q25.32, which is the long (q) arm of chromosome 3 at position 25.32

Molecular Location: base pairs 158,644,527 to 158,695,581 on chromosome 3 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EFG
- EFG1
- EFGM
- EGF1
- G translation elongation factor, mitochondrial
- GFM
- hEFG1
- mitochondrial elongation factor G
- mitochondrial elongation factor G1

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience: Molecular Biology of the OXPHOS System https://www.ncbi.nlm.nih.gov/books/NBK6292/
- The Cell: A Molecular Approach (second edition, 2000): The Genetic System of Mitochondria https://www.ncbi.nlm.nih.gov/books/NBK9896/#A1629

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28GFM1%5BTIAB%5D%29+OR+%28EFG1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- MITOCHONDRIAL ELONGATION FACTOR G1
  http://omim.org/entry/606639

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GFM1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GFM1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:13780
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:85476
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/85476
- UniProt
  https://www.uniprot.org/uniprot/Q96RP9

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